

# SNiPacard

## Block annotations

### Block info

genomic range	chr22:17,616,715-17,618,164 <i>e!</i>
block size	1,450 bp
variant count	3 variants

### Basic features

Conservation/deleteriousness		Linked genes	
phyloP	$\mu = -0.697$ [-1.53 – -0.194]	gene(s) hit or close-by	AC006946.15 <i>e!</i> , CECR5 <i>e!</i>
phastCons	$\mu = 0.002$ [0 – 0.005]	eQTL gene(s)	CECR6 <i>e!</i>
GERP++	$\mu = -0.386$ [-2.83 – 1.56]	potentially regulated gene(s)	-
CADD score	$\mu = 4.606$ [3.14 – 5.743]	disease gene(s)	-

## Direct effect on regulation

### cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CECR6 <i>e!</i>	?	ENSG00000183307 <i>e!</i>	sun exposed skin	4.38×10 <sup>-8</sup> (p-value)	GTEx Portal V6 <i>mq</i>	1

## Putative effect on regulation

### Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC006946.15 <i>e!</i>	downstream gene variant	3721	ENST00000441544 <i>e!</i>	?	?	1
CECR5 <i>e!</i>	downstream gene variant	237	ENST00000155674 <i>e!</i>	NM_017829.5	ENSP00000155674 <i>e!</i>	3
CECR5 <i>e!</i>	downstream gene variant	248	ENST00000477157 <i>e!</i>	?	?	3
CECR5 <i>e!</i>	downstream gene variant	308	ENST00000486462 <i>e!</i>	?	?	3
CECR5 <i>e!</i>	downstream gene variant	305	ENST00000399852 <i>e!</i>	?	ENSP00000382745 <i>e!</i>	3
CECR5 <i>e!</i>	downstream gene variant	246	ENST00000336737 <i>e!</i>	NM_033070.2	ENSP00000337358 <i>e!</i>	3

